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KLIPPEL – TRENAUNAY – WEBER SYNDROME WITH HEMIMEGALENCEPHALY*

Report of a Case

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SUMMARY: Alpay F, Kürekçi AE, Güneşli S, Gökçay E. (Department of Pediatrics, Gülhane Military Medical Academy, Ankara, Turkey). Klippel-Trenaunay-Weber syndrome with hemimegalencephaly: report of a case. Turk J Pediatr 1996; 38: 277-280.

A case of Klippel-Trenaunay-Weber Syndrome (KTWS) in a 2.5-year-old girl with congenital hemihypertrophy, nevus flammeus and liver hemangioma is presented. In addition, this report describes the rare association of hemimegalencephaly with KTWS.
Key words: hemihypertrophy, nevus flammeus, Klippel-Trenaunay-Weber syndrome, hemimegalencephaly.

The syndrome of a macular vascular nevus (nevus flammeus), bony and soft tissue hypertrophy, and venous varicosities was described in 1900 by Klippel and Trenaunay[1]. Weber[2] added another component, arteriovenous fistula, in 1907. These findings are also present in the clinical presentations of several other syndromes (Beckwith-Wiedeman, Silver-Russel, Sturge-Weber, Rubinstein-Taybi, Cobbs, etc.) and malignancies (Wilms' tumor, hepatoblastoma, etc.)[3,4]. Hemimegalencephaly, or unilateral megalencephaly, is caused by abnormal neuronal proliferation and migration during the second trimester, and results in overgrowth and secondary polymicrogyria of one cerebral hemisphere. The association of this entity with Klippel-Trenaunay-Weber syndrome (KTWS) is extremely rare. Cerebral and cerebellar hemihypertrophy have been reported previously[5], but magnetic resonance imaging (MRI) was not done in that case. We present a case of KTWS and her striking MRI findings with hemimegalencephaly.

Case Report

A 2.5-year-old girl was admitted to Gülhane Military Medical Academy Hospital with the findings of anemia, cervical lymphadenitis, macrosomic appearance, hemihypertrophy involving the left upper and lower extremities, nevus flammeus

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on the trunk, face, upper right extremity and lower extremities, hypopigmented skin lesions, and mental and motor subnormality (Fig. 1). Her height was at the 50th, weight at the 90th, chest circumference at the 75th, and head circumference above the 97th percentile. The hemoglobin was 4.5 g/dl, hematocrit 13 percent, platelet count 434,000/mm^3, and red cell indices revealed iron deficiency anemia [mean corpuscular volume (MCV) 56 cubic microns, mean corpuscular hemoglobin (MCH) 22 pg/cell, red-cell distribution width (RDW) 39.4]. The serum iron was 21 µg/dl, total iron binding capacity 342 µg/dl and ferritin 46 ng/ml, and occult blood was negative in the stool. Plain skeletal films showed cranial hemihypertrophy. The bone age was delayed (6 months-1 year). Bone scintigraphy confirmed the presence of soft tissue hemihypertrophy. Total body venography was normal except for left vesicoureteral reflux. Abdominal ultrasonography revealed diffuse enlargement of the liver, and computed tomography showed hemangioma in the liver (Fig. 2). Magnetic resonance imaging of the brain revealed dysmyelinization, left lateral ventricle dilatation. (particularly in the frontal and occipital horns), and hemimegalencephaly (Fig. 3). Skin biopsy verified the diagnosis of nevus flammeus, and skin fibroblast culture showed no mosaicism.

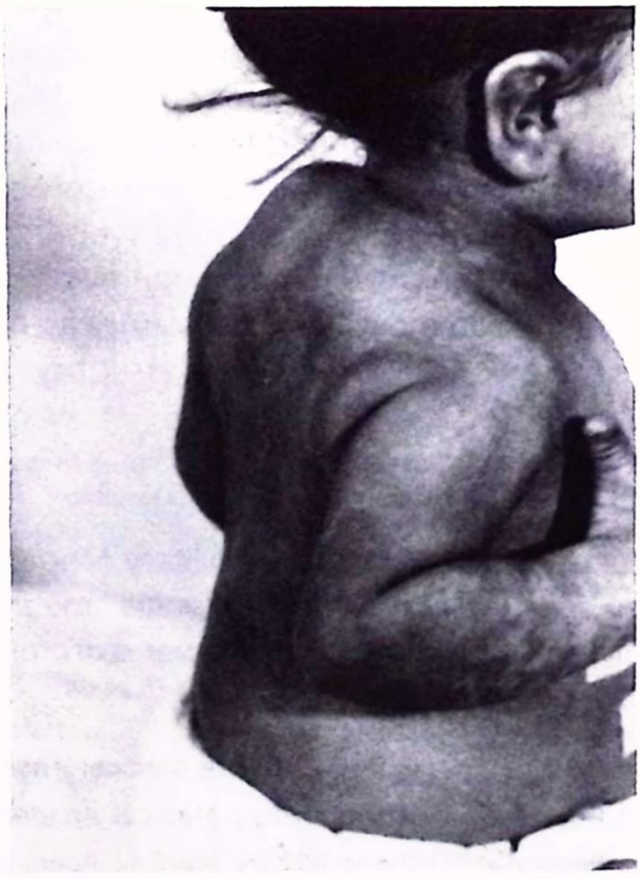

Fig. 1: Nevus flammeus on the trunk and arm.

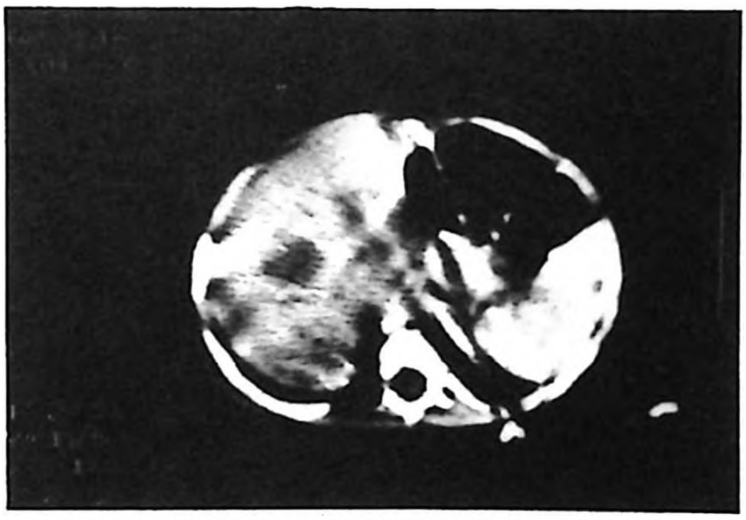

Fig. 2: Computed tomographic appearance of liver hemangioma.

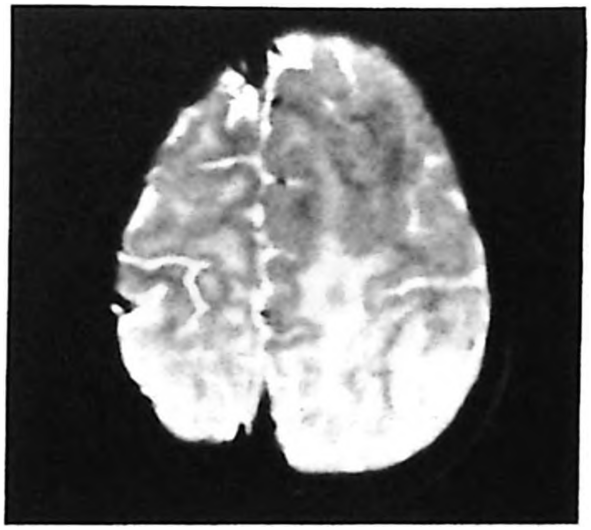

Fig. 3: Magnetic resonance imaging of hemimegalencephaly.

Discussion

Klippel-Trenaunay-Weber Syndrome consists of a port-wine nevus in combination with bony and soft tissue hypertrophy and venous varicosities. The association of hemimegalencephaly, hypopigmented skin lesions and the absence of bony hypertrophy are uncommon in this nonheritable disorder. Although some of its features resemble those of Hypomelanosis of Ito (i.e., hypopigmented macules, mental retardation, and craniofacial dysmorphism), the lack of chromosomal abnormality, scalp abnormalities, triphalangeal thumbs, inflammatory cells in the dermis, and pigment incontinence distinguish it from KTWS[6,7]. Several syndromes may mimic the clinical and laboratory features of KTWS. Of these, Maffucci syndrome has enchondromas in long bones[8], and Bannayan syndrome has

symmetrical macrocephaly, lipomatosis and angiomatosis[9] in association with hemihypertrophy. The lack of a porencephalic cyst with cerebral atrophy seen in Haberlund encephalocraniocutaneous lipomatosis and the absence of exostoses with characteristic cerebriform masses involving the palmar or plantar surfaces in Proteus syndrome exclude these syndromes in the differential diagnosis[10,11]. Cranial hemihypertrophy is a relatively rare component of KTWS[12,13]. Therefore, we believe that our patient is an interesting instance of KTWS exhibiting many unusual manifestations.

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